

## Abstract

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# Germline Mutations in BRCA1 and BRCA2 in Polish Families Predisposed to Breast and Ovary Cancers

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Germline mutations in BRCA1 and BRCA2 were analyzed in DNA isolated from white blood cells of patients with a family history of breast and ovarian cancers. We found one mutation in the BRCA1 gene — 300T/G missense mutation in exon 5 disrupting the zinc finger — already found in other families. In 5 families three different mutations were discovered in BRCA2. All these mutations are new and have not been described earlier. A frameshift mutation 9630delC causing truncation of the BRCA2 protein was identified in three different families which indicates a founder effect. Two other mutations were 9599A/T missense and 6886del5 frameshift, truncating the BRCA2 protein.



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